

Exon-Intron Boundary Sequences of the Human alpha-7 nAChR Subunit Gene

Exon Number	Exon length (bp)	cDNA position	splice acceptor	flanking exon sequence	splice donor	Intron number	Intron Size approximate (Kb)
1	55	1-55		...CTG CAC G L H	GTAAAGCCAC	1	0.3
2	140	56-195	TCTCCTTAAG	TG TCC.....GAC GTG V S D V	GTGAGTCCCG	2	Unknown
3	45	196-240	TTTTTTGAAG	GAT GAG.....CAA ATG D E Q M	GTAAGTTAAG	3	9.0
4	110	241-350	TGTGTGTCAG	TCT TGG.....AAC AG S W N S	GTAAGCATAT	4	Unknown
5	80	351-430	CTGTTTCTAG	T GCT GAT.....CCT CCA G A D P P	GTAAGCTGCA	5	4.0
6	168	431-598	ACCCACACAG	GC ATA.....CTA GTG G G I L V	GTAAGCCATG	6	1.0
7	195	599-793	CCCTATGGAG	GA ATC.....TCC CTG G G I S L	GTAAGCGCCC	7	1.0
8	87	794-880	TATGTTTTAG	GG ATA.....TTG ATA G G I L I	GTAAGGCAAG	8	3.5
9	110	881-990	CTCTCCACAG	CC CAG.....AAG TGG A Q K W	GTACGTTCTT	9	5.0
10	519	991-1509	GTCTCCCCCAG	ACC AGA... T R			

FIG. 1

Sequence Variants Identified in Full-Length and Duplicated Genomic Clones

DNA	EXONS CONT.	EXON 6 +/- 497-498	EXON 7 654	EXON 7 690	EXON 10 1269	EXON 10 1335	L76630
CHR15 HYBRID	5-10 1-10	+TG -TG	C/T	G/A	C/C		6GT 8GT
YAC							
D-948a10	5-10	-TG	T	A	C		6GT
D-853b12	6-10	-TG	T	A	C		6GT
D/F 969b11	5-10 1-10	+TG -TG	C/T	G/A	C/T		6GT 8GT
F-134h10	1-10	+TG	C		C		8GT
F-776a12	1-10	+TG	C		C		8GT
F-791e6	1-10	+TG	C		C		8GT
F-811b6	1-10	+TG	C		C		8GT
F-953g6	1-10	+TG	C		C		8GT
F-859c11	1-10	+TG	C		C		8GT
F-810f11	1-10	+TG	C		C		8GT
F-801e1	1-10	+TG	C		C		8GT
BAC							
F-467o18	1-10	+TG	C	G	C	T	8GT

FIG. 2

DNA	Control #	EXON 6 +/- 497-498			EXON 7 654			EXON 7 690			EXON 10 1269				EXON 10 1335		
Control Genomic DNA	43	+/+	+/-	-/-	C/C	C/T	T/T	G/G	G/A	A/A	C/C	C/T	T/T	C/C	C/T	T/T	
		10	33	0	5	38	0	0	43	0	6	36	1	24	19	0	

Expression Analysis of Sequence Variants

Subj	Bases 497-498			Base 654			Base 690			Base 933			Base 1296			Base 1335		
	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA
SL061	+/-	+	+/-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	C	C	C
SL084	+	+	+	C	C	C	GA	G	GA	G	G	G	CT	C	CT	C	C	C
SL111	+/-	+	+/-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	CT	CT	CT
SL097	+	+	+	CT	C	CT	GA	G	GA	G	G	G	CT	C	CT	C	C	C
SL089	+	+	+	C	C	C	GA	GA	GA	GA	GA	GA	CT	CT	CT	C	C	C
SHSY	+/-	+	+/-	CT	C	CT	GA	GA	GA	GA	GA	GA	C	C	C	C	C	C

FIG. 3

-392 agaacgcaag ggagaggtag agcctggcct tgggcagccc ctggdctggc cagaggcgcg aggccgagag
AP-2

-322 ccgcctcggt ggagactggg ggtggaggtg ccgggagcgt acccagegcc gggagtacct cccgctcaca

-252 cctcgggctg cagtccctg ggtggccgcc gagacgctgg cccgggctgg agggatggcg gggcggggac

-182 gggggcgggg gcggggctcg tcacptggag aggcgcggcg gggcggggcg gccgcgccgcg
CREB Sp1

-112 tccttaagg cgcgcgagcc ggcggcgag gtgccttgt ggcgcgagcc gcaggccccg gcgacagcg

-42 agacgtggag cgcgccggct cgctgcagct ccgggactca acATGCGCTG CTCGCCGGGA GCGCTCTGCG
Met

+29 TGGCCTGCG CCGTCCCTC CTGCACGGTA aagccac

CENTROMERE		ALPHA-7 SEQUENCE										ALPHA-7 SEQUENCE										TELOMERE			
PAC BAC YAC	SIZE kb	D15S1043	D15S942	EXON 5	EXON 6	EXON 7	EXON 8	EXON 9	L76630	EXON 10	L76630	EXON 9	EXON 8	EXON 7	EXON 6	EXON 5	EXON 4	EXON 3	EXON 2	EXON 1	D15S1360	D15S1010	D15S144	D15S1007	D15S995
PAC		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
64a1		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
25919		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
BAC		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
467 18		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
YAC		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
948a10	1730	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-	-
853b12	790	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-	-
895f6	1580	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
969b11	1030	-	-	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-
776a12	1640	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
791e6	1170	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
811b6	1060	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
953g6	1720	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
134h10	N.A.	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
859c11	1330	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
810f11	940	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
801e1	1630	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
966a4	1500	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
764f8	740	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-
822g2	1280	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-

FIG. 5

EXON D 297bp	1 CAGGCCGCCA CAGCGCGCG AGCGGCTCGG TTCAAGGCCA CCCACCGCAA	CATAGCTCCC GGGAGAGGTG GACTGGCTCC AACCGAAGTT CATTAAGgtg 297	GCCAAgTCCT GAGCCGGGAG TTTTCCGGCG ACTGGCCTCT agtcgcc.....	CGGTGCCCTT AGCTCGGCCG CCCTCCCGCC ATCTTCCAGG agtcgcc.....	TGCCATTTTC GGGGCCCCCG GGAGGTGAGG AGAACCCAGGA agtcgcc.....	CAGCCGCGTC CTGGTGGCCG GGAAGATGTC GCCACAGCCG agtcgcc.....	CCACGAGGGT CGGCCATGAC CATGTCAGGG CGGCTCACGC agtcgcc.....
EXON C 125bpctc TATCTACACG AAGtgagttg 422	atttcagATT ACTCAGATCT ta.....	ACAAGTGGAC TGTTGTCACC ta.....	ACCTGAGTCA CCCATTATTG ta.....	GCAGGACCTG ACAATCCAAA ta.....	GAATCCCAGA GGTGCAGAAA ta.....	TGAGAGAGCT GCACTCTGAC ta.....
EXON B 64bp	...ttaaccac TTTCAGgtag 486	agATAATGAA gatcat.....	ACAACCACCA ta.....	TGGGTTAAAT ta.....	TTGATGCAAA ta.....	AATATTGCAT ta.....	CTACCAGCAT ta.....
EXON A 47bpttta aca.....	ttctagTTCC ta.....	AATTGCTAAT ta.....	CCAGCATTTG ta.....	TGGATAGCTG ta.....	CAAAC TGCGA 533	TATgtaagta 533
EXON 5 80bp	...ctgtttc ta.....	tagTGCTGAT GCATTGCCAG 614	GAGCGCTTTG TACCTGCCTC ta.....	ACGCCACATT CAGgtaagctgca..... 613	CCACACTAAC ta.....	GTGTTGGTGA ta.....	ATTCTTCTGG ta.....
EXON 6 27bpaccacac ta.....	cagGCATATT ta.....	CAAGAGTTCC ta.....	TGCTACATCG 640	ta.....	ta.....	ta.....

FIG. 6

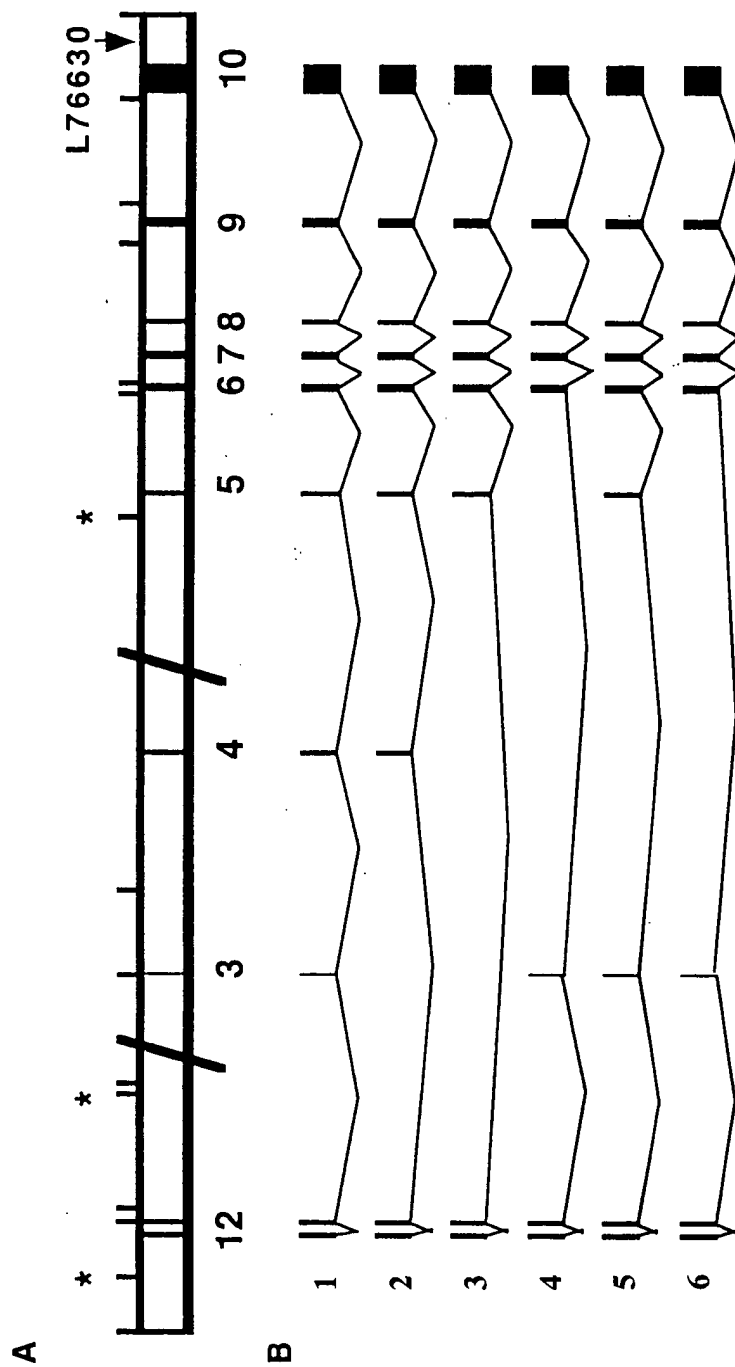


FIG. 7

```

1  agaacgcaag ggagaggtag agcctggcct tgggcagccc ctggcctggc cagaggcgcg
61  aggccgagag cccgctcggt ggagactggg ggtggaggtg cccggagcgt acccagcgcc
121  gggagtacct cccgctcaca cctcgggctg cagttccctg ggtggccgcc gagacgtgg
181  cccgggctgg agggatggcg gggcggggac gggggcgggg gcggggctcg tcacgtggag
241  aggcgcgcgg gggcggggcg ggcggggggc gcgccccggc tccttaaagg cgcgcgagcc
301  gagcggcgag gtgcctctgt ggccgcaggc gcaggcccg gcgacagccg agacgtggag
361  cgcgccggct cgctgcagct ccgggactca ac

```

FIG. 8

```

1  caggccgcca catagctccc gccaaagtcc cggtgcccct tgccattttc cagccgcgct
61  cccacgaggg tcacggcggc ggggagaggt ggagccgcga gagctcggcc gggggccccc
121  cctggtggcc gcggccatga cagcggctcg ggactggctc cttttccgcg cccctcccg
181  cggaggtgag gggaagatgt ccatgtcagg gttcaaggcc aaaccgaagt tactggcctc
241  tatcttccag gagaaccagg agccacagcc gcggctcacg cccaccgca acattaagat
301  tacaagtgga cacctgagtc agcaggacct ggaatcccag atgagagagc ttatctacac
361  gactcagatc ttgttgtcac cccattatt gacaatccaa aggtgcagaa agcactctga
421  caaataatga aacaaccacc atcgggttaa tttgatgcaa aaatattgca tctaccagca
481  ttttcagttc caattgctaa tccagcattt gtggatagct gcaaactgcg atattgctga
541  tgagcgcttt gacgccacat tccacactaa cgtgttggtg aattcttctg ggcattgcca
601  gtacctgcct ccaggcatat tcaagagttc ctgctacatc g

```

FIG. 9

```

1  agccctttcc caggcggtag cgggggcagt ggtgctgttg cccttttaaa ctgcggcttg
61  acgggagccg cgcctcctgt cggtggagtc ggttataaag ggagcagccc cgcaggccgc
121  cacatagctc ccgccaagtc ctcggtgccc cttgccattt tccagccgcy ctcaccgag
181  ggtcacggcg gcggggagag gtggagccgc gagagctcgg ccggggggccc cgcctggtgg
241  ccgcggccat gacagcggct cgggactggc tccttttccg cgccttccc gccggaggtg
301  aggggaagat gtccatgtca gggttcaagg ccaaaccgaa gttactggcc tctatcttcc
361  aggagaacca ggagccacag ccgcggctca cgcgccaccg caacattaag attacaagtg
421  gacacctgag tcagcaggac ctggaatccc agatgagaga gcttatctac acgactcaga
481  tcttggtgtc acccccatta ttgacaatcc aaagggtgcag aaagcactct gacaattcca
541  attgctaate cagcatttgt ggatagctgc aaactgcgat attgctgatg agcgctttga
601  cgccacattc cactactaac tgttggtgaa ttcttctggg cattgccagt acctgcctcc
661  aggcattatc aagagttcct gctacatcg

```

FIG. 10

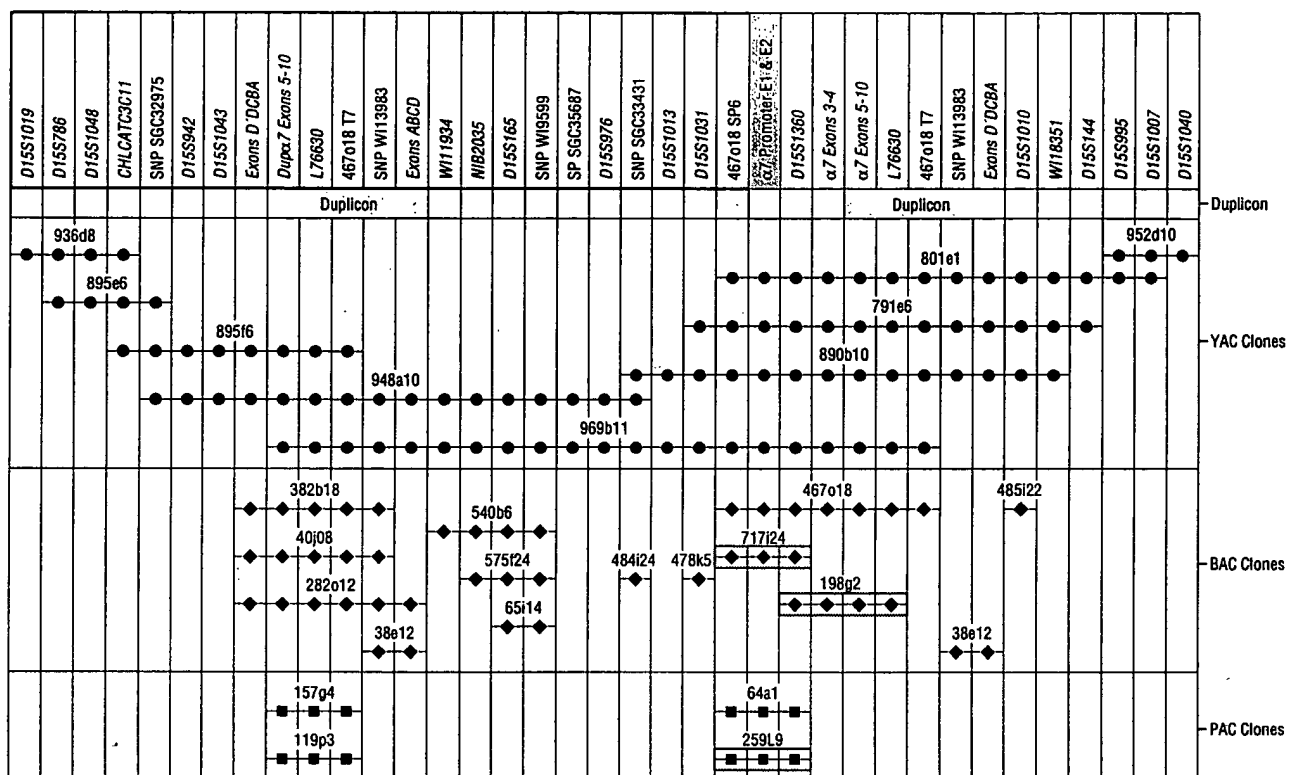


FIG. 11

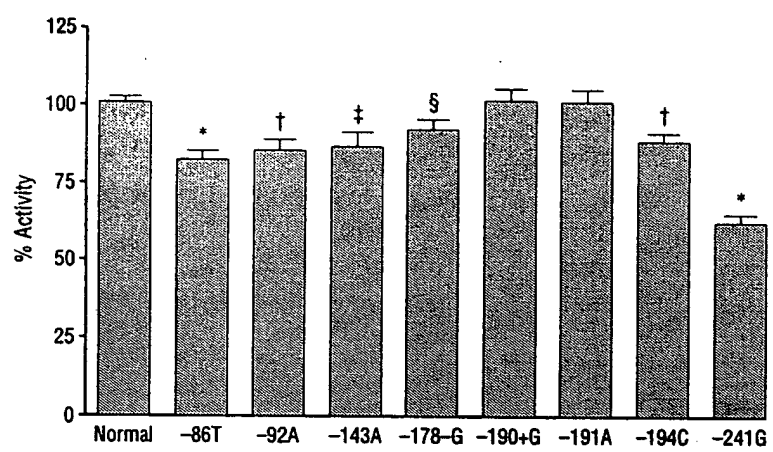


FIG. 13

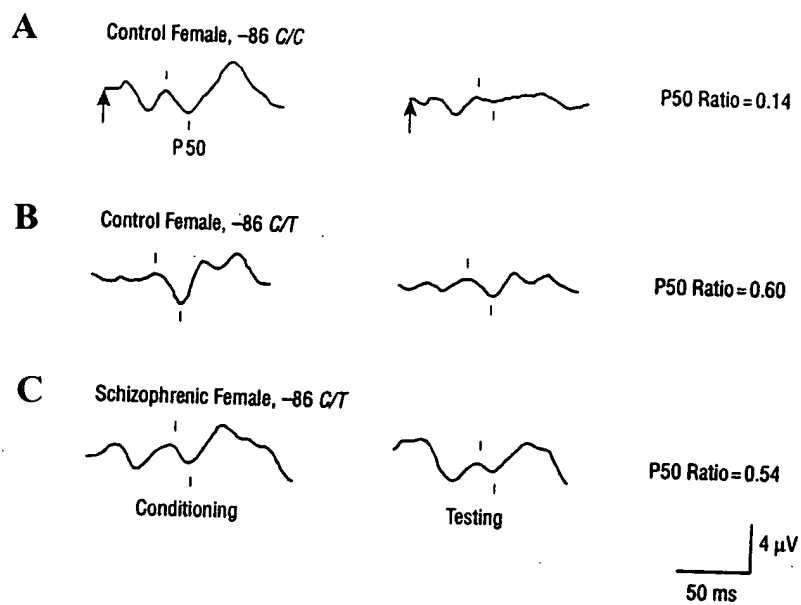
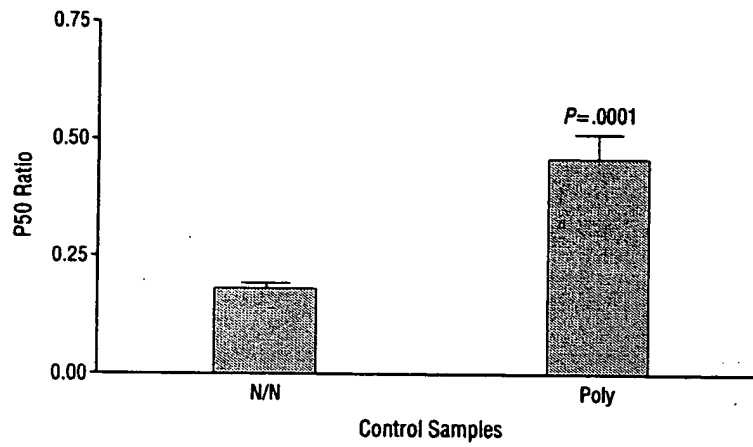


FIG. 14

A



B

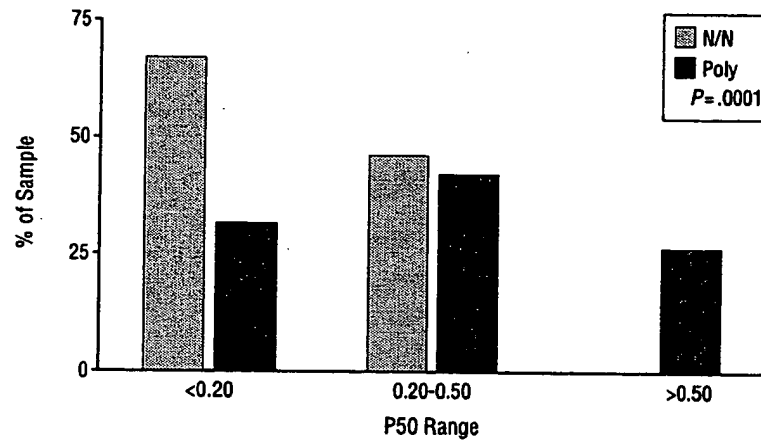
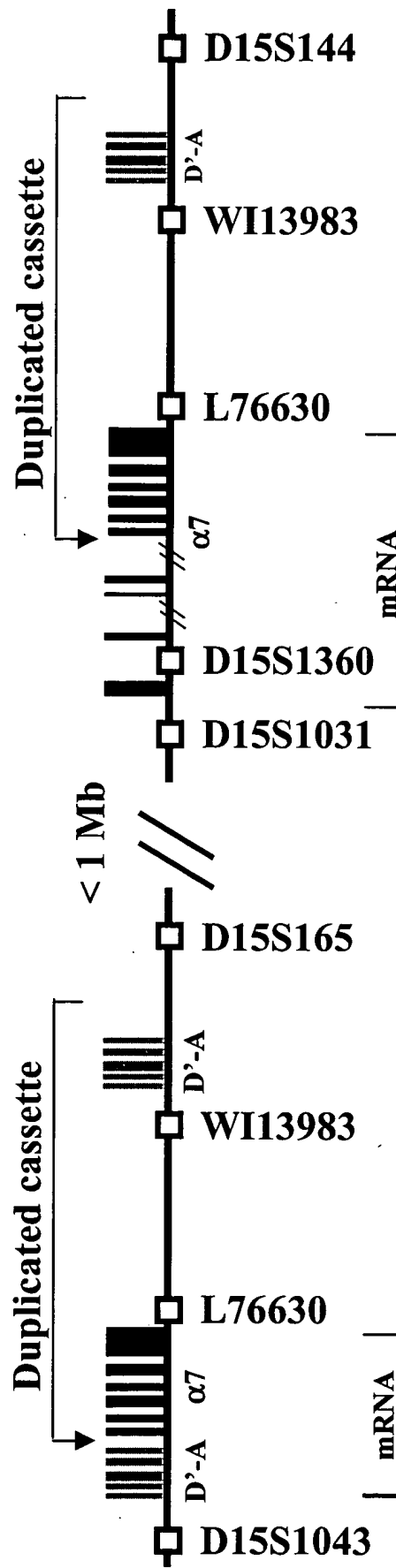


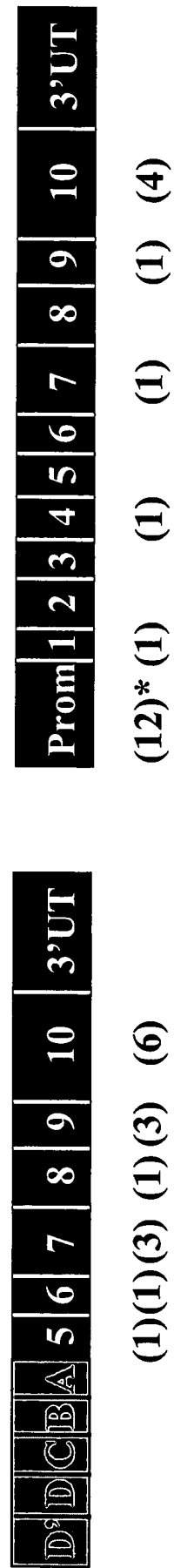
FIG. 15

FIG. 16

a



b



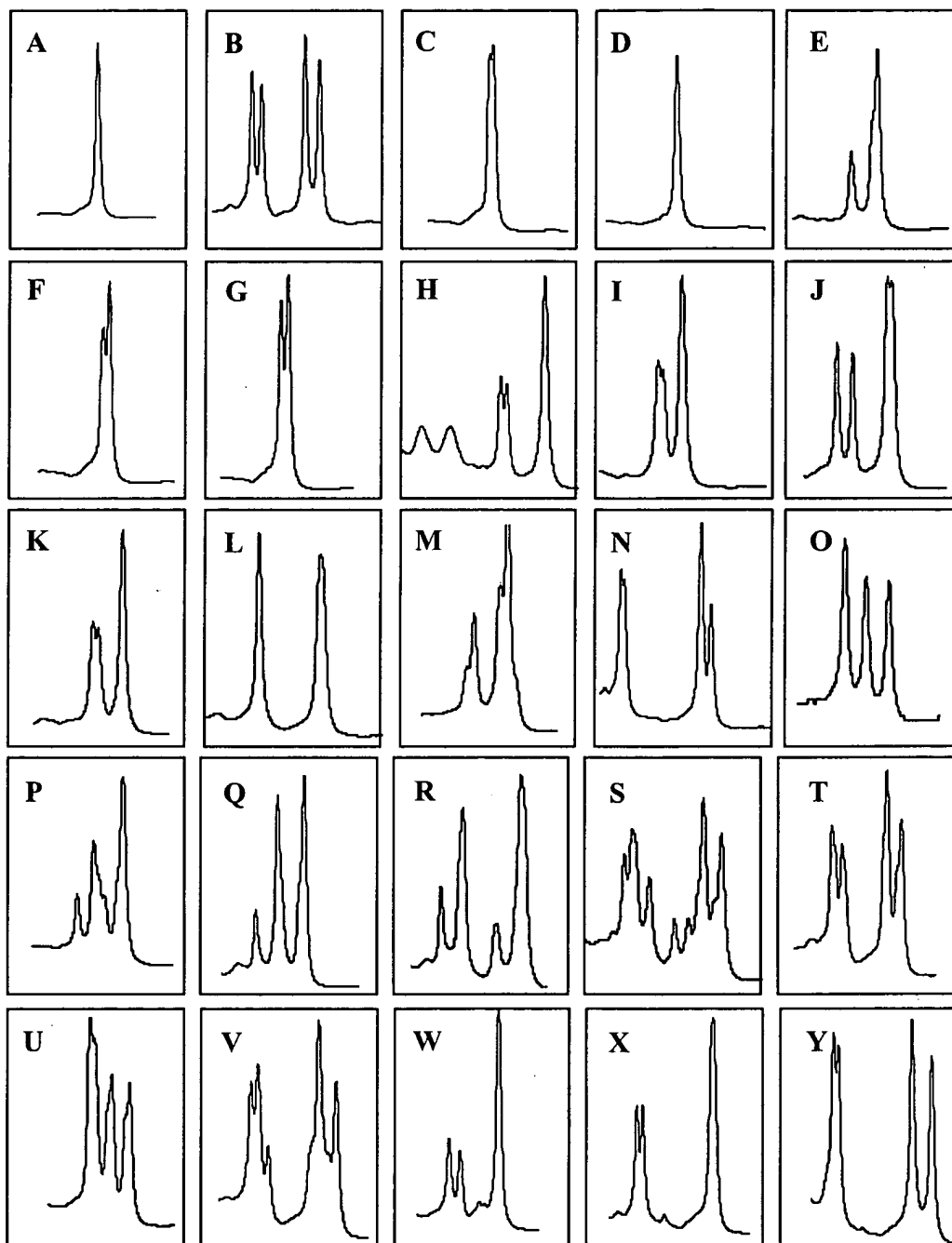


Fig. 17